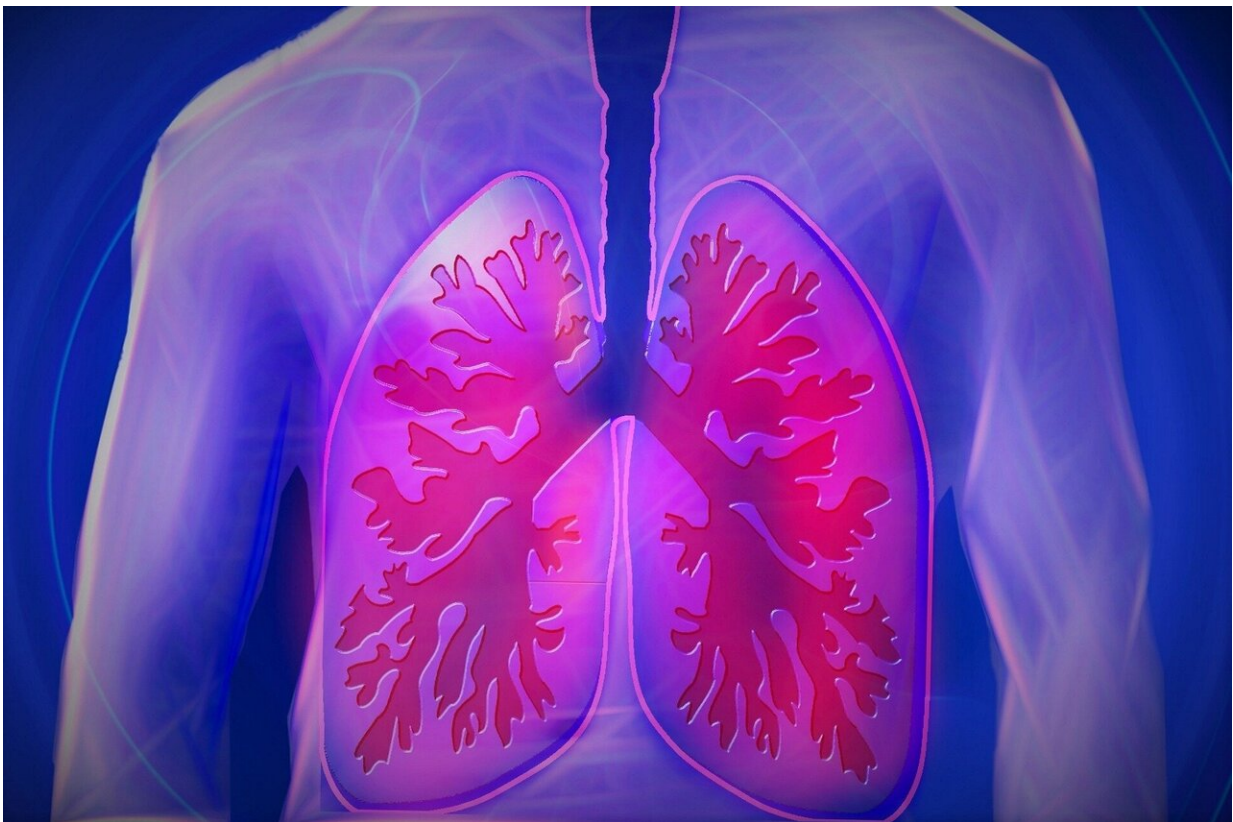


Diagnosis of genetic condition could help patients stop smoking and prevent lung disease

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New research shows that people diagnosed with a genetic condition, called alpha-1 antitrypsin deficiency (AATD), are far more likely to stop

smoking and therefore prevent the development of lung disease.

The study, led by researchers from RCSI University of Medicine and Health Science, is published in *COPD: Journal of Chronic Obstructive Pulmonary Disease*.

It is estimated that 265,000 people on the island of Ireland are affected by either severe or moderate AATD, but the vast majority of people with AATD have not been diagnosed.

Previously, it was assumed that only people with severe AATD were at risk of [lung](#) disease. Recent Irish research has shown that people with the far more common moderate form of AATD are also at risk of lung disease if they are smokers.

With the [smoking rate](#) in the [general population](#) in Ireland at 17%, this means there are approximately 45,000 current smokers with either severe or moderate AATD who are at increased risk of lung damage due to cigarettes. According to the latest figures from the national targeted detection program for AATD based at RCSI Beaumont Hospital, 90% of these smokers with AATD remain undiagnosed.

The researchers surveyed patients enrolled in the National AATD Registry with a questionnaire relating to demographics, parental smoking history, personal smoking history, AATD awareness and personal medical history.

Of the 293 respondents, 58 reported being smokers at the time of their AATD diagnosis. Their subsequent reported quit rate was 70.7%.

"Our study has shown that those who receive a diagnosis of alpha-1 antitrypsin deficiency are far more likely to stop smoking. We hope this study will lead to increased testing for AATD, with more people being

diagnosed and choosing to stop or completely avoid [smoking](#) in order to prevent lung [disease](#) from developing," said Dr. Tomás Carroll, senior lecturer at RCSI and the study's corresponding author.

"People with AATD who use electronic cigarettes are also potentially at risk, but more research is needed."

The research also found that people who reported having a parent who smoked were far more likely to become [smokers](#) themselves. Those with a parent who smoked were 84% more likely to smoke than those who did not.

For a diagnosis of AATD, a simple blood test is available in most large hospitals in Ireland that can identify those who need further testing. The more precise definitive test is provided for free by Alpha-1 Foundation Ireland as part of a HSE funded national targeted detection program.

More information: Alessandro N. Franciosi et al, Alpha-1 Antitrypsin Deficiency and Tobacco Smoking: Exploring Risk Factors and Smoking Cessation in a Registry Population, *COPD: Journal of Chronic Obstructive Pulmonary Disease* (2021). [DOI: 10.1080/15412555.2020.1864725](#)

Provided by RCSI

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